



South Carolina Department of Health  
and Environmental Control

# NEWBORN SCREENING

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## How long has DHEC done this testing?

Testing for phenylketonuria (PKU) began in 1965. A 1976 law made testing mandatory unless parents had religious objections. The other tests were recommended by the Newborn Screening Advisory Committee and approved by the DHEC board.

## What diseases are screened?

*Phenylketonuria* is an error in amino acid metabolism resulting in profound mental retardation. The consequences can be avoided by very early detection and a diet low in the amino acid phenylalanine. About three cases a year are detected.

*Congenital Hypothyroidism* results from the lack of thyroid hormone leading to mental retardation and slow growth. If detected early, thyroid hormone can be given and the consequences reduced or eliminated. About 10 babies are born each year with primary congenital hypothyroidism.

*Hemoglobinopathy (Sickle Cell Disease)* patients may have impaired oxygen transfer ability and may have many painful and life-threatening conditions, some of which can be prevented through early diagnosis and monitoring. About 100 babies a year are born with hemoglobinopathy disorders including sickle cell disease while another 2,400 are born as carriers.

*Galactosemia* is caused by an inability to metabolize galactose, a component of lactose found in milk and other foods. Serious gastrointestinal disturbances, failure to gain weight and jaundice may occur. Children with untreated galactosemia develop mental retardation, cataracts and speech delay. Some infants die in the early neonatal period from bacterial sepsis. If detected early, diets low in galactose can minimize the effects of the condition. About one baby a year is born with this condition.

*Congenital adrenal hyperplasia* is an enzyme defect that affects the functioning of the adrenal gland with the overproduction of testosterone. If untreated, females are virilized (made masculine). Children with CAH may begin puberty at 3-5 years of age. In some cases, the child may experience dehydration, shock and death. Administration of adrenal hormones can prevent some serious consequences. About three babies a year are born with this condition.

*Medium chain acyl-CoA dehydrogenase deficiency* causes an error in fatty acid metabolism resulting in a child's blood sugar becoming dangerously low. About 20 percent of children with MCADD die before diagnosis if screening is not in place and those who survive may have serious residual effects. If detected early, the condition can be treated by making sure the patient gets enough carbohydrate, especially during illness or other stressful times. About four babies a year are born with this condition.

**Do parents sign a consent form?**

There is no DHEC consent form for newborn screening. The DHEC Newborn Screening Manual; Law, Regulations, and Official Departmental Instructions states, "The provision of the screening test is covered under the informed consent signed by the parents at the hospital." Parents may refuse the tests on religious objections.

**Do parents get any information in advance?**

The regulations require hospital staff to provide information about the testing. The hospital may give the parents a pamphlet with information about the testing.

**How long does it take to do the testing?**

On average, testing is completed three working days after the specimen arrives in DHEC's laboratory. If the results indicate a high likelihood that the infant has a newborn screening disorder, the physician of record is notified by phone by staff in the Division of Women and Children's Services. If the results are less definite but still abnormal, the physician will be notified by mail. The laboratory needs to keep the specimens, which are blood spots, long enough to complete testing and allow for requests for routine additional tests or repeats. A period of six months would be adequate for these needs.

**What happens to the information?**

Results are sent to the hospital or mid-wife and to the physician of record. Abnormal results are immediately sent to DHEC's Division of Women and Children's Services for follow-up. The results from children with sickle-cell trait (carriers of sickle cell) are sent by the Division of Women and Children's Services to regional sickle-cell centers for counseling and follow-up.

**Why does DHEC keep the cards?**

Since 1994, state law required the cards be stored, frozen, for an unspecified time. Prior to that, they were kept on a retention schedule of originally 25 years to allow the child to reach adulthood and to allow for a period of legal discovery should there be any lawsuits that might involve the tests. In 1988, the retention schedule was changed to 10 years. We also must comply with federal licensing (CLIA) regulations, which directs that all test request forms be kept for at least two years. In the case of the newborn screening, the blood spot specimen is an integral part of the request form and, has been stored.

**When did DHEC begin keeping the cards?**

We began about 1976.

**Where does DHEC store the cards?**

Since 1995, cards have been stored in the Hayne Building, in a locked freezer. The cards before 1995 were stored at the S.C. Department of Archives and History.

**Who has access to the information?**

In DHEC's Hayne Building, only staff members with direct access to the freezer. At the Department of Archives, their procedures are followed with only limited access allowed. The cards are stored in numbered containers with no patient-identifying information visible.

**Why are there cards at State Archives?**

The forms needed to be in a large, secure facility. In 1976, DHEC did not have a secure warehouse that could handle some 55,000 forms each year. The Department of Archives had the warehouse space and agreed to accept the forms and keep them on a retention schedule of 25 years. That time frame was changed to 10 years in 1988. The forms that are stored at the Department of Archives can be released only to DHEC.

**Does DHEC do any DNA-based newborn screening or store purified DNA?**

No. DHEC does no DNA-based (gene) newborn screening. The DNA is stored only because DNA is present in the blood cells. No purified DNA is stored. The information from the blood spots can be released only to the parents of the child, the child's doctor and to the child, once he or she reaches the age of 18.